



SBF2 gene

SET binding factor 2

Normal Function

The *SBF2* gene (also called *MTMR13*) provides instructions for making a protein called SET binding factor 2. The function of this protein is unknown, but it is probably involved in the development of specialized cells in the nervous system called Schwann cells. Schwann cells produce myelin, the protective substance that covers nerve cells and promotes the rapid transmission of nerve impulses. SET binding factor 2 probably also plays a role in the development of mesh-like canals (trabecular meshwork) that surround the colored part of the eye (the iris). The trabecular meshwork helps drain excess fluid from the eye.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

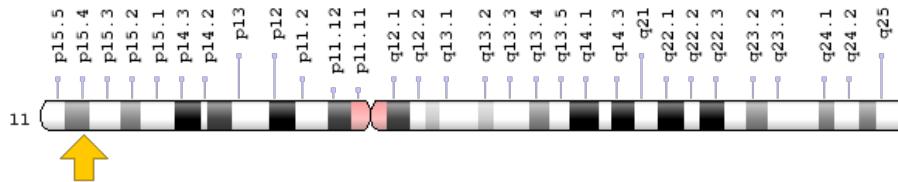
At least five *SBF2* gene mutations have been identified in patients with a form of Charcot-Marie-Tooth disease known as type 4B2. Some of these mutations alter the structure of SET binding factor 2 by introducing a premature stop signal that results in an abnormally short protein. Other mutations lead to the production of a protein that is missing a critical segment. All of these mutations probably result in a nonfunctional protein. Although it is unclear how *SBF2* gene mutations lead to this disorder, myelin production is probably disrupted. Irregular myelin structure (called outfolding) is a characteristic sign of type 4B2 Charcot-Marie-Tooth disease.

Individuals with this disorder may also experience a buildup of fluid pressure within the eye (glaucoma) beginning in childhood or adolescence. Researchers believe that the appearance of glaucoma depends on the type of *SBF2* gene mutation. A mutation that causes complete loss of protein function interferes with the development of the eye's trabecular meshwork, leading to impaired fluid drainage and glaucoma. Less severe mutations, which allow partial function of the SET binding factor 2 protein, do not cause glaucoma.

Chromosomal Location

Cytogenetic Location: 11p15.4, which is the short (p) arm of chromosome 11 at position 15.4

Molecular Location: base pairs 9,778,667 to 10,294,216 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT4B2
- DENND7B
- KIAA1766
- MTMR13
- MTMRD_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Myelin facilitates conduction
<https://www.ncbi.nlm.nih.gov/books/NBK27954/#A245>

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4
<https://www.ncbi.nlm.nih.gov/books/NBK1468>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CMT4B2%5BTIAB%5D%29+OR+%28%28MTMR13%5BTIAB%5D%29+OR+%28SBF2%5BTIAB%5D%29+OR+%28SET+binding+factor+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SET-BINDING FACTOR 2
<http://omim.org/entry/607697>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SBF2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SBF2%5Bgene%5D>
- HGNC Gene Family: DENN/MADD domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/504>
- HGNC Gene Family: Myotubularins
<http://www.genenames.org/cgi-bin/genefamilies/set/903>
- HGNC Gene Family: Pleckstrin homology domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/682>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2135
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=24>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/81846>
- UniProt
<http://www.uniprot.org/uniprot/Q86WG5>

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